

1. INTRODUCTION & BACKGROUND

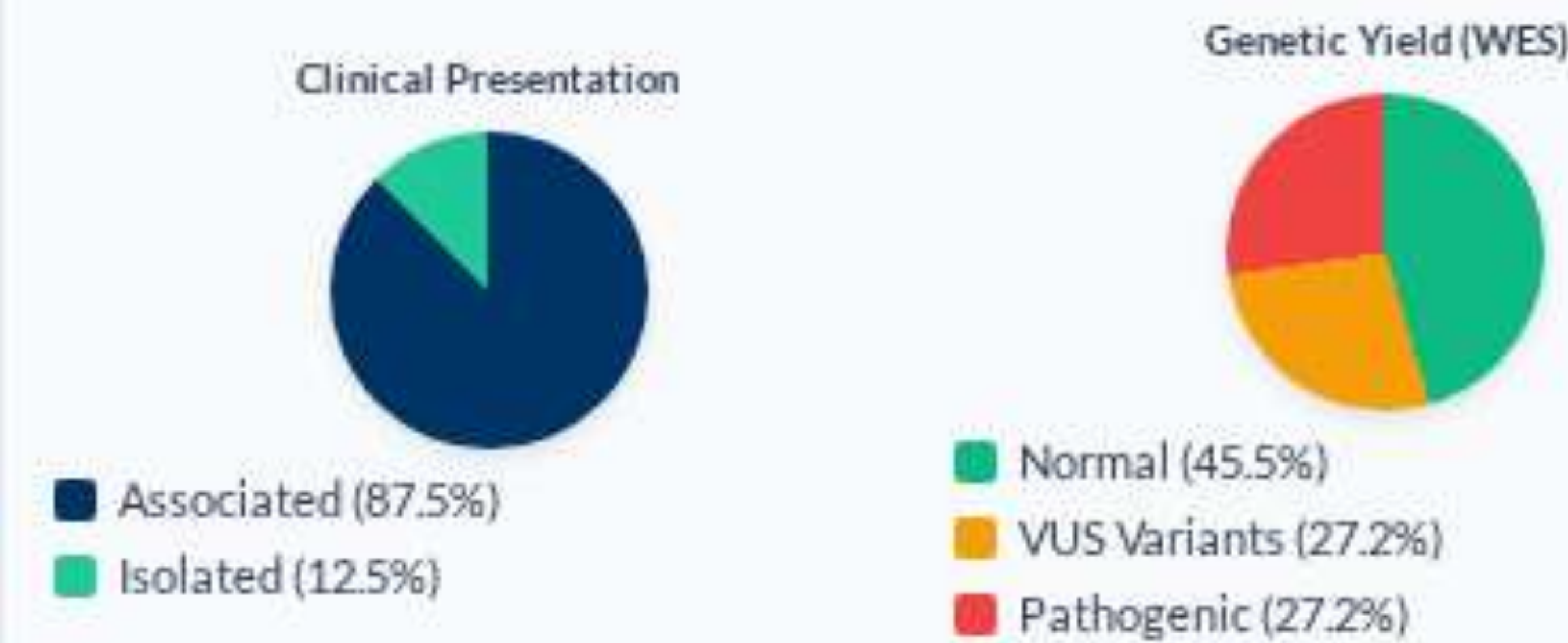
Binder syndrome (maxillonasal dysplasia) is traditionally identified by local midfacial hypoplasia, a flattened nasal bridge, and an increased nasofrontal angle. However, emerging diagnostic evidence suggests that this phenotype represents a much broader, under-recognized multisystem pathology.

Primary Objective: Retrospective review of 16 prenatal cases to characterize fetal anomalies, map genetic etiologies, and evaluate pregnancy outcomes.

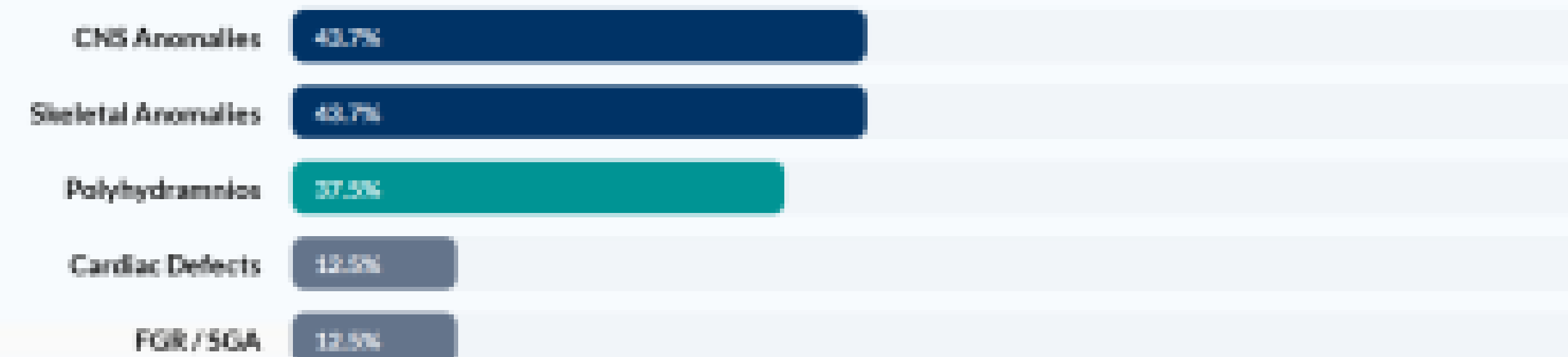
2. METHODOLOGY FLOW DIAGRAM

- Screening Cohort:** All singleton pregnancies scanned between 2023 and 2025 showing signs of midfacial hypoplasia.
- Anatomical Evaluation:** Profile and facial assessments using *GE Voluson E10* & *Philips EPIQ Elite*.
- Multisystem Scan:** Comprehensive fetal anatomical survey highlighting CNS, cardiac, and skeletal matrices.
- Exome Sequencing:** Targeted prenatal Whole Exome Sequencing (WES) conducted in 11/16 cases (68.7%).

3. RESULTS & VISUAL PROPORTIONS



Spectrum of Associated Abnormalities



4. MAP OF IDENTIFIED PATHOGENIC GENES

Targeted WES revealed severe genetic heterogeneity behind the Binder phenotype. Critical gene pathologies identified include:

COL18A1

Knobloch syndrome. Encodes collagen XVIII $\alpha 1$, causing eye and brain defects.

ARSL

CDPX1. X-linked recessive disorder altering bone and cartilage development.

SOX9

Campomelic Dysplasia. Vital transcription factor regulating chondrogenesis.

5. ULTRASOUND PHENOTYPING



Figure 1: High-res midsagittal fetal profile scan demonstrating depressed nasal bridge and verticalized nasal bone.

6. PREGNANCY OUTCOMES

Clinical management was heavily influenced by genetic outcomes and multisystem comorbidities:

62.5%

Live Birth (10/16)

37.5%

Termination (6/16)

★REFINED CONCLUSION

"Binder syndrome is a critical sentinel marker of severe genetic and multisystem pathology rather than an isolated facial variant. Identification of this phenotype demands early targeted exome sequencing and holistic fetal mapping to drive accurate clinical and parental counseling."